MAYO CLINIC LABORATORIES

NGHHA and Subpanel Comparison Gene List

Gene		NGHHA	NGENZ	NGMEM	NGCDA	Associated Disorders
AK1	Adenylate kinase	X	X			Adenylate kinase deficiency
ALDOA	Aldolase	X	X			Aldolase deficiency
ANK1	Ankyrin	X		X		Hereditary Spherocytosis
CDIN1	CDAN1 interacting nuclease 1	X			X	Congenital Dyserythropoietic Anemia (CDA) type Ib
CD59	MAC-IP, MIRL, protectin	X				*CD59*-mediated hemolytic anemia with immune-mediated polyneuropathy)/Neonatal anemia
CDAN1	Codanin1	X			X	Congenital Dyserythropoietic Anemia (CDA) type Ia
EPB41	Protein 4.1	X		X		Hereditary Elliptocytosis, Pyropoikilocytosis
EPB42	Protein 4.2	X		X		Hereditary Spherocytosis
FANCA	Fanconi anemia, complementation group A	X				Fanconi anemia
FANCC	Fanconi anemia, complementation group C	X				Fanconi anemia
FANCG	Fanconi anemia, complementation group G	X				Fanconi anemia
G6PD	Glucose 6 phosphate dehydrogenase	X	X			G6PD deficiency
GATA1	*GATA* binding protein 1	X			X	X-linked thrombocytopenia with thalassemia/ Congenital Dyserythropoietic Anemia (CDA) variant
GCLC	Gamma Glutamate Cysteine Ligase	X	X			Glutathione deficiency
GPI	Glucose 6 Phosphate isomerase	X	X			GPI deficiency
GSR	Glutathione Reductase	X	X			Glutathione deficiency
GSS	Glutathione Synthetase	X	X			Glutathione deficiency
GYPC	Glycophorin C and D	X		X		Hereditary elliptocytosis
HBB	Beta globin	X	X	X	X	Beta chain hemoglobin variants and thalassemia
HBD	Delta globin	X	X	X	X	Delta chain hemoglobin variants and thalassemia
HK1	Hexokinase	X	X			HK deficiency
HMOX1	Heme Oxygenase 1	X	X			HMOX1 deficiency (hemolysis, Fe accumulation, growth retardation), Increased Hb F
KIF23	Kinesin family member	X			X	Congenital Dyserythropoietic Anemia (CDA) type III
KLF1	Kruppel-like factor 1	X			X	Congenital Dyserythropoietic Anemia (CDA) type IV, increased Hb A2, non-deletional HPFH
NT5C3A	5-Nucleotidase cytosolic 3A	X	X			P5NT deficiency (Pyrimidine 5' nucleotidase/uridine 5' monophosphate hydrolase UMPH1 deficiency)
PFKM	Phosphofructokinase	X	X			PFK deficiency (glycogen storage disease VII/ Tarui disease)
PGK1	Phosphoglycerate Kinase	X	X			PGK1 deficiency
PIEZO1	PIEZO ion channel	X		X		Dehydrated Hereditary Stomatocytosis/ Hereditary Xerocytosis, perinatal edema
PKLR	Pyruvate Kinase	X	X			PK deficiency
RHAG	Rhesus Blood Group Associated Glycoprotein	X		X		Overhydrated Hereditary Stomatocytosis, Rh-null hemolytic anemia

NGHHA and Subpanel Comparison Gene List (continued)

Gene		NGHHA	NGENZ	NGMEM	NGCDA	Associated Disorders
RPS19	Ribosomal Protein S19	X				Diamond-Blackfan anemia
SEC23B	*SEC23* homolog B, coat complex II component	X			X	Congenital Dyserythropoietic Anemia (CDA) type II
SLC2A1	Solute Carrier Family 2 (GLUT1)	X		X		Stomatin-deficient Cryohydrocytosis
SLC4A1	Solute Carrier Family 4 (Band3)	X		X		Hereditary Spherocytosis, SE Asian Ovalocytosis, Cryohydrocytosis, stomatocytosis, acanthocytosis, distal renal tubular acidosis with hemolysis
SPTA1	Spectrin, alpha	X		X		Hereditary Spherocytosis, Hereditary Elliptocytosis, Pyropoikilocytosis
SPTB	Spectrin, beta	X		X		Hereditary Spherocytosis, Hereditary Elliptocytosis, Pyropoikilocytosis
STOM	Stomatin (Protein 7.2)	X		X		Stomatocytosis
TPI1	Triosephosphate isomerase	X	X			TPI deficiency
UGT1A1	UDP Glycosyltransferase 1 A1	X	X	X	X	Hyperbilirubinemia, Gilbert syndrome, Crigler-Najjar syndrome type I and II